

SEQUENCE LISTING

IAP13 Rec'd PCT/PTO 09 DEC 2005

<110> Pietrangelo, Antonello

<120> Mutations in the SLC40A1 gene associated to impaired iron homeostasis

<130> 8907-109-999

<140> To be Determined

<141> 2005-12-09

<150> PCT/EP2004/051068

<151> 2004-06-09

<160> 30

<170> PatentIn version 3.1

<210> 1

<211> 1716

<212> DNA

<213> Homo sapiens

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<221> CDS

<222> (1)..(1716)

<223> cDNA encoding wild type ferroportin 1. Polymorphisms related to the codons:
238-240 (G80), 520-522 (N174), 742-744 (Q248)

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tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg	144
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val	
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Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr	
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Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly	
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gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg	288
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu	
85 90 95	
gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg	336
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met	
100 105 110	
gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt	384

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Val	Val	Ala	Gly	Glu	Asp	Arg	Ser	Lys	Leu	Ala	Asn	Met	Asn	Ala	Thr		
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ata	cga	agg	att	gac	cag	tta	acc	aac	atc	tta	gcc	ccc	atg	gct	gtt		576
Ile	Arg	Arg	Ile	Asp	Gln	Leu	Thr	Asn	Ile	Leu	Ala	Pro	Met	Ala	Val		
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Gly	Gln	Ile	Met	Thr	Phe	Gly	Ser	Pro	Val	Ile	Gly	Cys	Gly	Phe	Ile		
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Lys	Val	Tyr	Gln	Lys	Thr	Pro	Ala	Leu	Ala	Val	Lys	Ala	Gly	Leu	Lys		
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gaa	gag	gaa	act	gaa	ttg	aaa	cag	ctg	aat	tta	cac	aaa	gat	act	gag		768
Glu	Glu	Glu	Thr	Glu	Leu	Lys	Gln	Leu	Asn	Leu	His	Lys	Asp	Thr	Glu		
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cca	aaa	ccc	ctg	gag	gga	act	cat	cta	atg	ggt	gtg	aaa	gac	tct	aac		816
Pro	Lys	Pro	Leu	Glu	Gly	Thr	His	Leu	Met	Gly	Val	Lys	Asp	Ser	Asn		
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cag	cct	gtg	ttt	ctg	gct	ggc	atg	ggt	ctt	gct	ttc	ctt	tat	atg	act		960
Gln	Pro	Val	Phe	Leu	Ala	Gly	Met	Gly	Leu	Ala	Phe	Leu	Tyr	Met	Thr		
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gct aga atc ggt ctt tgg tcc ttt gat tta act gtg aca cag ttg ctg Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu 465 470 475 480			1440
caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln 485 490 495			1488
aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile 500 505 510			1536
ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val 515 520 525			1584
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act ctg gga aac aag ctc ttt gct tgc ggt cct gat gca aaa gaa gtt Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val 545 550 555 560			1680
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35 40 45
Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
50 55 60
Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
65 70 75 80
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
85 90 95
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
100 105 110
Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val
115 120 125
Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn
130 135 140
Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val
145 150 155 160
Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr
165 170 175
Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val
180 185 190
Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile
195 200 205
Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp
210 215 220
Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys
225 230 235 240

Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu
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 Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn
 260 265 270
 Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met
 275 280 285
 Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn
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 Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr
 305 310 315 320
 Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly
 325 330 335
 Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr
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 Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly
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 Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu
 370 375 380
 Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu
 385 390 395 400
 Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu
 405 410 415
 Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met
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 Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu
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 Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu
 465 470 475 480
 Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln

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Asn Ser Met	Asn Tyr Leu Leu Asp	Leu Leu His Phe Ile Met Val Ile			
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Leu Ala Pro	Asn Pro Glu Ala Phe Gly Leu Leu Val	Leu Ile Ser Val			
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Ser Phe Val	Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn				
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 <223> cDNA encoding a ferroportin 1 mutated in position (G80).

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Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His	
20 25 30	
tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg	144
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val	
35 40 45	
ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac	192
Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr	
50 55 60	
ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc agt	240
Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Ser	
65 70 75 80	
gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg	288
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu	
85 90 95	
gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg	336
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met	

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Val	Phe	Leu	His	Lys	His	Glu	Leu	Leu	Thr	Met	Tyr	His	Gly	Trp	Val	
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ctc	act	tcc	tgc	tat	atc	ctg	atc	atc	act	att	gca	aat	att	gca	aat	432
Leu	Thr	Ser	Cys	Tyr	Ile	Leu	Ile	Ile	Thr	Ile	Ala	Asn	Ile	Ala	Asn	
	130					135					140					
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Val	Val	Ala	Gly	Glu	Asp	Arg	Ser	Lys	Leu	Ala	Asn	Met	Asn	Ala	Thr	
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Ile	Arg	Arg	Ile	Asp	Gln	Leu	Thr	Asn	Ile	Leu	Ala	Pro	Met	Ala	Val	
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ggc	cag	att	atg	aca	ttt	ggc	tcc	cca	gtc	atc	ggc	tgt	ggc	ttt	att	624
Gly	Gln	Ile	Met	Thr	Phe	Gly	Ser	Pro	Val	Ile	Gly	Cys	Gly	Phe	Ile	
	195						200					205				
tcg	gga	tgg	aac	ttg	gta	tcc	atg	tgc	gtg	gag	tac	gtc	ctg	ctc	tgg	672
Ser	Gly	Trp	Asn	Leu	Val	Ser	Met	Cys	Val	Glu	Tyr	Val	Leu	Leu	Trp	
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Lys	Val	Tyr	Gln	Lys	Thr	Pro	Ala	Leu	Ala	Val	Lys	Ala	Gly	Leu	Lys	
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gaa	gag	gaa	act	gaa	ttg	aaa	cag	ctg	aat	tta	cac	aaa	gat	act	gag	768
Glu	Glu	Glu	Thr	Glu	Leu	Lys	Gln	Leu	Asn	Leu	His	Lys	Asp	Thr	Glu	
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cca	aaa	ccc	ctg	gag	gga	act	cat	cta	atg	ggt	gtg	aaa	gac	tct	aac	816
Pro	Lys	Pro	Leu	Glu	Gly	Thr	His	Leu	Met	Gly	Val	Lys	Asp	Ser	Asn	
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gct	gag	ccc	ttc	cgt	acc	ttc	cga	gat	gga	tgg	gtc	tcc	tac	tac	aac	912
Ala	Glu	Pro	Phe	Arg	Thr	Phe	Arg	Asp	Gly	Trp	Val	Ser	Tyr	Tyr	Asn	
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cag	cct	gtg	ttt	ctg	gct	ggc	atg	ggt	ctt	gct	ttc	ctt	tat	atg	act	960
Gln	Pro	Val	Phe	Leu	Ala	Gly	Met	Gly	Leu	Ala	Phe	Leu	Tyr	Met	Thr	
305					310					315					320	
gtc	ctg	ggc	ttt	gac	tgc	atc	acc	aca	ggg	tac	gcc	tac	act	cag	gga	1008
Val	Leu	Gly	Phe	Asp	Cys	Ile	Thr	Thr	Gly	Tyr	Ala	Tyr	Thr	Gln	Gly	
				325					330					335		
ctg	agt	ggt	tcc	atc	ctc	agt	att	ttg	atg	gga	gca	tca	gct	ata	act	1056
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Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu	
370 375 380	
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Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu	
385 390 395 400	
tcc gtt tct cct ttt gaa gat atc cga tca agg ttc att caa gga gag	1248
Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu	
405 410 415	
tca att aca cct acc aag ata cct gaa att aca act gaa ata tac atg	1296
Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met	
420 425 430	
tct aat ggg tct aat tct gct aat att gtc ccg gag aca agt cct gaa	1344
Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu	
435 440 445	
tct gtg ccc ata atc tct gtc agt ctg ctg ttt gca ggc gtc att gct	1392
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450 455 460	
gct aga atc ggt ctt tgg tcc ttt gat tta act gtg aca cag ttg ctg	1440
Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu	
465 470 475 480	
caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag	1488
Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln	
485 490 495	
aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc	1536
Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile	
500 505 510	
ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc	1584
Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val	
515 520 525	
tcc ttt gtg gca atg ggc cac att atg tat ttc cga ttt gcc caa aat	1632
Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn	
530 535 540	
act ctg gga aac aag ctc ttt gct tgc ggt cct gat gca aaa gaa gtt	1680
Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val	
545 550 555 560	
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Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val	
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35 40 45

Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
50 55 60

Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Ser
65 70 75 80

Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
85 90 95

Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
100 105 110

Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val
115 120 125

Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn
130 135 140

Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val
145 150 155 160

Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr
165 170 175

Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val
180 185 190

Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile
195 200 205

Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp
210 215 220

Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys

225		230		235		240									
Glu	Glu	Glu	Thr	Glu	Leu	Lys	Gln	Leu	Asn	Leu	His	Lys	Asp	Thr	Glu
				245					250					255	
Pro	Lys	Pro	Leu	Glu	Gly	Thr	His	Leu	Met	Gly	Val	Lys	Asp	Ser	Asn
			260					265					270		
Ile	His	Glu	Leu	Glu	His	Glu	Gln	Glu	Pro	Thr	Cys	Ala	Ser	Gln	Met
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Ala	Glu	Pro	Phe	Arg	Thr	Phe	Arg	Asp	Gly	Trp	Val	Ser	Tyr	Tyr	Asn
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Gln	Pro	Val	Phe	Leu	Ala	Gly	Met	Gly	Leu	Ala	Phe	Leu	Tyr	Met	Thr
305					310					315					320
Val	Leu	Gly	Phe	Asp	Cys	Ile	Thr	Thr	Gly	Tyr	Ala	Tyr	Thr	Gln	Gly
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Leu	Ser	Gly	Ser	Ile	Leu	Ser	Ile	Leu	Met	Gly	Ala	Ser	Ala	Ile	Thr
			340					345					350		
Gly	Ile	Met	Gly	Thr	Val	Ala	Phe	Thr	Trp	Leu	Arg	Arg	Lys	Cys	Gly
		355					360					365			
Leu	Val	Arg	Thr	Gly	Leu	Ile	Ser	Gly	Leu	Ala	Gln	Leu	Ser	Cys	Leu
	370					375					380				
Ile	Leu	Cys	Val	Ile	Ser	Val	Phe	Met	Pro	Gly	Ser	Pro	Leu	Asp	Leu
385					390					395					400
Ser	Val	Ser	Pro	Phe	Glu	Asp	Ile	Arg	Ser	Arg	Phe	Ile	Gln	Gly	Glu
				405					410					415	
Ser	Ile	Thr	Pro	Thr	Lys	Ile	Pro	Glu	Ile	Thr	Thr	Glu	Ile	Tyr	Met
			420					425					430		
Ser	Asn	Gly	Ser	Asn	Ser	Ala	Asn	Ile	Val	Pro	Glu	Thr	Ser	Pro	Glu
		435					440					445			
Ser	Val	Pro	Ile	Ile	Ser	Val	Ser	Leu	Leu	Phe	Ala	Gly	Val	Ile	Ala
	450					455					460				
Ala	Arg	Ile	Gly	Leu	Trp	Ser	Phe	Asp	Leu	Thr	Val	Thr	Gln	Leu	Leu
465					470					475					480

Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln
485 490 495

Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile
500 505 510

Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val
515 520 525

Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn
530 535 540

Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val
545 550 555 560

Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val
565 570

<210> 5

<211> 1716

<212> DNA

<213> Homo sapiens

<220>

<221> CDS

<222> (1)..(1716)

<223> cDNAencoding a ferroportin 1 mutated in position 174 (N174)

<400> 5

atg acc agg gcg gga gat cac aac cgc cag aga gga tgc tgt gga tcc 48
Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser
1 5 10 15

ttg gcc gac tac ctg acc tct gca aaa ttc ctt ctc tac ctt ggt cat 96
Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
20 25 30

tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg 144
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
35 40 45

ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac 192
Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
50 55 60

ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc ggt 240
Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
65 70 75 80

gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg 288
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
85 90 95

gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg	336
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met	
100 105 110	
gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt	384
Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val	
115 120 125	
ctc act tcc tgc tat atc ctg atc atc act att gca aat att gca aat	432
Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn	
130 135 140	
ttg gcc agt act gct act gca atc aca atc caa agg gat tgg att gtt	480
Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val	
145 150 155 160	
gtt gtt gca gga gaa gac aga agc aaa cta gca aat atg att gcc aca	528
Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Ile Ala Thr	
165 170 175	
ata cga agg att gac cag tta acc aac atc tta gcc ccc atg gct gtt	576
Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val	
180 185 190	
ggc cag att atg aca ttt ggc tcc cca gtc atc ggc tgt ggc ttt att	624
Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile	
195 200 205	
tcg gga tgg aac ttg gta tcc atg tgc gtg gag tac gtc ctg ctc tgg	672
Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp	
210 215 220	
aag gtt tac cag aaa acc cca gct cta gct gtg aaa gct ggt ctt aaa	720
Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys	
225 230 235 240	
gaa gag gaa act gaa ttg aaa cag ctg aat tta cac aaa gat act gag	768
Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu	
245 250 255	
cca aaa ccc ctg gag gga act cat cta atg ggt gtg aaa gac tct aac	816
Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn	
260 265 270	
atc cat gag ctt gaa cat gag caa gag cct act tgt gcc tcc cag atg	864
Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met	
275 280 285	
gct gag ccc ttc cgt acc ttc cga gat gga tgg gtc tcc tac tac aac	912
Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn	
290 295 300	
cag cct gtg ttt ctg gct ggc atg ggt ctt gct ttc ctt tat atg act	960
Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr	
305 310 315 320	
gtc ctg ggc ttt gac tgc atc acc aca ggg tac gcc tac act cag gga	1008
Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly	
325 330 335	

ctg agt ggt tcc atc ctc agt att ttg atg gga gca tca gct ata act Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr 340 345 350	1056
gga ata atg gga act gta gct ttt act tgg cta cgt cga aaa tgt ggt Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly 355 360 365	1104
ttg gtt cgg aca ggt ctg atc tca gga ttg gca cag ctt tcc tgt ttg Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu 370 375 380	1152
atc ttg tgt gtg atc tct gta ttc atg cct gga agc ccc ctg gac ttg Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu 385 390 395 400	1200
tcc gtt tct cct ttt gaa gat atc cga tca agg ttc att caa gga gag Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu 405 410 415	1248
tca att aca cct acc aag ata cct gaa att aca act gaa ata tac atg Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met 420 425 430	1296
tct aat ggg tct aat tct gct aat att gtc ccg gag aca agt cct gaa Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu 435 440 445	1344
tct gtg ccc ata atc tct gtc agt ctg ctg ttt gca ggc gtc att gct Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala 450 455 460	1392
gct aga atc ggt ctt tgg tcc ttt gat tta act gtg aca cag ttg ctg Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu 465 470 475 480	1440
caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln 485 490 495	1488
aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile 500 505 510	1536
ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val 515 520 525	1584
tcc ttt gtg gca atg ggc cac att atg tat ttc cga ttt gcc caa aat Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn 530 535 540	1632
act ctg gga aac aag ctc ttt gct tgc ggt cct gat gca aaa gaa gtt Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val 545 550 555 560	1680
agg aag gaa aat caa gca aat aca tct gtt gtt tga Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val 565 570	1716

<210> 6
 <211> 571
 <212> PRT
 <213> Homo sapiens

<400> 6

Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser
 1 5 10 15

Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
 20 25 30

Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
 35 40 45

Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
 50 55 60

Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
 65 70 75 80

Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
 85 90 95

Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
 100 105 110

Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val
 115 120 125

Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn
 130 135 140

Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val
 145 150 155 160

Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Ile Ala Thr
 165 170 175

Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val
 180 185 190

Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile
 195 200 205

Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp
 210 215 220

Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys
 225 230 235 240

Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu
 245 250 255

Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn
 260 265 270

Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met
 275 280 285

Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn
 290 295 300

Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr
 305 310 315 320

Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly
 325 330 335

Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr
 340 345 350

Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly
 355 360 365

Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu
 370 375 380

Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu
 385 390 395 400

Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu
 405 410 415

Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met
 420 425 430

Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu
 435 440 445

Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala
 450 455 460

Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu
465 470 475 480

Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln
485 490 495

Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile
500 505 510

Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val
515 520 525

Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn
530 535 540

Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val
545 550 555 560

Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val
565 570

<210> 7
<211> 1716
<212> DNA
<213> Homo sapiens

<220>
<221> CDS
<222> (1)..(1716)
<223> cDNA encoding a ferroportina 1 mutated in position 248 (Q248).

<400> 7
atg acc agg gcg gga gat cac aac cgc cag aga gga tgc tgt gga tcc 48
Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser
1 5 10 15
ttg gcc gac tac ctg acc tct gca aaa ttc ctt ctc tac ctt ggt cat 96
Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
20 25 30
tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg 144
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
35 40 45
ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac 192
Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
50 55 60
ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc ggt 240
Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
65 70 75 80

gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg	288
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu	
85 90 95	
gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg	336
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met	
100 105 110	
gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt	384
Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val	
115 120 125	
ctc act tcc tgc tat atc ctg atc atc act att gca aat att gca aat	432
Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn	
130 135 140	
ttg gcc agt act gct act gca atc aca atc caa agg gat tgg att gtt	480
Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val	
145 150 155 160	
gtt gtt gca gga gaa gac aga agc aaa cta gca aat atg aat gcc aca	528
Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr	
165 170 175	
ata cga agg att gac cag tta acc aac atc tta gcc ccc atg gct gtt	576
Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val	
180 185 190	
ggc cag att atg aca ttt ggc tcc cca gtc atc ggc tgt ggc ttt att	624
Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile	
195 200 205	
tcg gga tgg aac ttg gta tcc atg tgc gtg gag tac gtc ctg ctc tgg	672
Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp	
210 215 220	
aag gtt tac cag aaa acc cca gct cta gct gtg aaa gct ggt ctt aaa	720
Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys	
225 230 235 240	
gaa gag gaa act gaa ttg aaa cat ctg aat tta cac aaa gat act gag	768
Glu Glu Glu Thr Glu Leu Lys His Leu Asn Leu His Lys Asp Thr Glu	
245 250 255	
cca aaa ccc ctg gag gga act cat cta atg ggt gtg aaa gac tct aac	816
Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn	
260 265 270	
atc cat gag ctt gaa cat gag caa gag cct act tgt gcc tcc cag atg	864
Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met	
275 280 285	
gct gag ccc ttc cgt acc ttc cga gat gga tgg gtc tcc tac tac aac	912
Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn	
290 295 300	
cag cct gtg ttt ctg gct ggc atg ggt ctt gct ttc ctt tat atg act	960
Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr	
305 310 315 320	
gtc ctg ggc ttt gac tgc atc acc aca ggg tac gcc tac act cag gga	1008

Val	Leu	Gly	Phe	Asp 325	Cys	Ile	Thr	Thr	Gly 330	Tyr	Ala	Tyr	Thr	Gln 335	Gly		
ctg	agt	ggg	tcc	atc	ctc	agt	att	ttg	atg	gga	gca	tca	gct	ata	act		1056
Leu	Ser	Gly	Ser	Ile	Leu	Ser	Ile	Leu	Met	Gly	Ala	Ser	Ala	Ile	Thr		
			340					345					350				
gga	ata	atg	gga	act	gta	gct	ttt	act	tg	cta	cgt	cga	aaa	tgt	ggg		1104
Gly	Ile	Met	Gly	Thr	Val	Ala	Phe	Thr	Trp	Leu	Arg	Arg	Lys	Cys	Gly		
		355					360					365					
ttg	gtt	cgg	aca	ggg	ctg	atc	tca	gga	ttg	gca	cag	ctt	tcc	tgt	ttg		1152
Leu	Val	Arg	Thr	Gly	Leu	Ile	Ser	Gly	Leu	Ala	Gln	Leu	Ser	Cys	Leu		
	370					375					380						
atc	ttg	tgt	gtg	atc	tct	gta	ttc	atg	cct	gga	agc	ccc	ctg	gac	ttg		1200
Ile	Leu	Cys	Val	Ile	Ser	Val	Phe	Met	Pro	Gly	Ser	Pro	Leu	Asp	Leu		
385					390					395					400		
tcc	gtt	tct	cct	ttt	gaa	gat	atc	cga	tca	agg	ttc	att	caa	gga	gag		1248
Ser	Val	Ser	Pro	Phe	Glu	Asp	Ile	Arg	Ser	Arg	Phe	Ile	Gln	Gly	Glu		
				405					410					415			
tca	att	aca	cct	acc	aag	ata	cct	gaa	att	aca	act	gaa	ata	tac	atg		1296
Ser	Ile	Thr	Pro	Thr	Lys	Ile	Pro	Glu	Ile	Thr	Thr	Glu	Ile	Tyr	Met		
			420					425					430				
tct	aat	ggg	tct	aat	tct	gct	aat	att	gtc	ccg	gag	aca	agt	cct	gaa		1344
Ser	Asn	Gly	Ser	Asn	Ser	Ala	Asn	Ile	Val	Pro	Glu	Thr	Ser	Pro	Glu		
		435					440					445					
tct	gtg	ccc	ata	atc	tct	gtc	agt	ctg	ctg	ttt	gca	ggc	gtc	att	gct		1392
Ser	Val	Pro	Ile	Ile	Ser	Val	Ser	Leu	Leu	Phe	Ala	Gly	Val	Ile	Ala		
	450					455					460						
gct	aga	atc	ggg	ctt	tg	tcc	ttt	gat	tta	act	gtg	aca	cag	ttg	ctg		1440
Ala	Arg	Ile	Gly	Leu	Trp	Ser	Phe	Asp	Leu	Thr	Val	Thr	Gln	Leu	Leu		
465					470				475						480		
caa	gaa	aat	gta	att	gaa	tct	gaa	aga	ggc	att	ata	aat	ggg	gta	cag		1488
Gln	Glu	Asn	Val	Ile	Glu	Ser	Glu	Arg	Gly	Ile	Ile	Asn	Gly	Val	Gln		
			485						490					495			
aac	tcc	atg	aac	tat	ctt	ctt	gat	ctt	ctg	cat	ttc	atc	atg	gtc	atc		1536
Asn	Ser	Met	Asn	Tyr	Leu	Leu	Asp	Leu	Leu	His	Phe	Ile	Met	Val	Ile		
			500					505					510				
ctg	gct	cca	aat	cct	gaa	gct	ttt	ggc	ttg	ctc	gta	ttg	att	tca	gtc		1584
Leu	Ala	Pro	Asn	Pro	Glu	Ala	Phe	Gly	Leu	Leu	Val	Leu	Ile	Ser	Val		
		515					520					525					
tcc	ttt	gtg	gca	atg	ggc	cac	att	atg	tat	ttc	cga	ttt	gcc	caa	aat		1632
Ser	Phe	Val	Ala	Met	Gly	His	Ile	Met	Tyr	Phe	Arg	Phe	Ala	Gln	Asn		
	530					535					540						
act	ctg	gga	aac	aag	ctc	ttt	gct	tgc	ggg	cct	gat	gca	aaa	gaa	gtt		1680
Thr	Leu	Gly	Asn	Lys	Leu	Phe	Ala	Cys	Gly	Pro	Asp	Ala	Lys	Glu	Val		
545					550				555						560		
agg	aag	gaa	aat	caa	gca	aat	aca	tct	gtt	gtt	tga						1716
Arg	Lys	Glu	Asn	Gln	Ala	Asn	Thr	Ser	Val	Val							

565

570

<210> 8

<211> 571

<212> PRT

<213> Homo sapiens

<400> 8

Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser
 1 5 10 15

Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
 20 25 30

Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
 35 40 45

Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
 50 55 60

Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
 65 70 75 80

Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
 85 90 95

Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
 100 105 110

Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val
 115 120 125

Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn
 130 135 140

Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val
 145 150 155 160

Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr
 165 170 175

Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val
 180 185 190

Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile
 195 200 205

Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp
 210 215 220
 Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys
 225 230 235 240
 Glu Glu Glu Thr Glu Leu Lys His Leu Asn Leu His Lys Asp Thr Glu
 245 250 255
 Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn
 260 265 270
 Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met
 275 280 285
 Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn
 290 295 300
 Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr
 305 310 315
 Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly
 325 330 335
 Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr
 340 345 350
 Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly
 355 360 365
 Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu
 370 375 380
 Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu
 385 390 395 400
 Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu
 405 410 415
 Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met
 420 425 430
 Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu
 435 440 445

Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala
 450 455 460

Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu
 465 470 475 480

Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln
 485 490 495

Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile
 500 505 510

Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val
 515 520 525

Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn
 530 535 540

Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val
 545 550 555 560

Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val
 565 570

<210> 9
 <211> 20
 <212> DNA
 <213> Homo sapiens

<220>
 <221> polymerase chain reaction primer
 <222> (1)..(20)
 <223> 5' PCR primer. Exon 1

<400> 9
 ggtgctatct ccagttcctt 20

<210> 10
 <211> 20
 <212> DNA
 <213> Homo sapiens

<220>
 <221> misc_feature
 <222> (1)..(20)
 <223> 3' PCR primer. Exon1

<400> 10
 gttcacagca gagccacatt 20

<210> 11
 <211> 25
 <212> DNA
 <213> Homo sapiens

 <220>
 <221> misc_feature
 <222> (1)..(25)
 <223> 5' PCR primer. Exon 2

<400> 11
 cagctcatta agtgactacc atcgc

25

<210> 12
 <211> 24
 <212> DNA
 <213> Homo sapiens

 <220>
 <221> misc_feature
 <222> (1)..(24)
 <223> 3' PCR primer. exon 2

<400> 12
 ggcttaatac aactggctag aacg

24

<210> 13
 <211> 23
 <212> DNA
 <213> Homo sapiens

 <220>
 <221> misc_feature
 <222> (1)..(23)
 <223> 5' PCR primer. Exon 3

<400> 13
 cataatgtag ccaggaagtg ccc

23

<210> 14
 <211> 22
 <212> DNA
 <213> Homo sapiens

 <220>
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 <222> (1)..(22)
 <223> 3' PCR primer. Exon 3

<400> 14
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22

<210> 15
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 <223> 5' PCR primer. Exon 4

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24

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 <223> 3' PCR primer. Exon 4

<400> 16
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24

<210> 17
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<400> 17
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24

<210> 18
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 <223> 3' PCR primer. Exon 5

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24

<210> 19

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 <400> 21
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 <210> 23
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<212> DNA
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 <222> (1)..(24)
 <223> 5' PCR primer. Exon 8

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 <210> 24
 <211> 24
 <212> DNA
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 <220>
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 <222> (1)..(24)
 <223> 3' PCR primer. Exon 8

 <400> 24
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 <210> 25
 <211> 10
 <212> DNA
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 <220>
 <221> misc_feature
 <222> (1)..(10)
 <223> oligonucleotide comprising the polymorphism at nt 238

 <400> 25
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 <210> 26
 <211> 10
 <212> DNA
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 <220>
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 <223> oligonucleotide comprising the polymorphism at nt 521.

 <400> 26
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 <210> 27
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 <212> DNA
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<222> (1)..(10)
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gaaacatctg

10

<210> 28
<211> 5
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<220>
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<223> X different from glycine

<400> 28

Ile Ile Xaa Asp Trp
1 5

<210> 29
<211> 5
<212> PRT
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<220>
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<222> (1)..(5)
<223> X different form asparagine

<400> 29

Asn Met Xaa Ala Thr
1 5

<210> 30
<211> 5
<212> PRT
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<220>
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<223> X different from glutamine

<400> 30

Leu Lys Xaa Leu Asn
1 5